

Opis choroby *

Definicja

A rare arthrogyposis syndrome characterized by arthrogyposis multiplex congenita with contractures involving multiple joints of the upper and lower limbs, camptodactyly of fingers and toes, skeletal abnormalities such as scoliosis and *pectus excavatum*, as well as variable speech and motor delay and hypotonia. Facial dysmorphism includes long eyelashes, periorbital fullness, ptosis, epicanthal folds, high arched/cleft palate, and micrognathia.

Dane

Klasyfikacja

Choroba

Synonimy

MYBPC1-related autosomal recessive non-lethal
AMC syndrome
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AMC syndrome

Kod ORPHA

498693

Kod OMIM

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Kod ICD10

Q74.3

Kod ICD11

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*Źródło

orphanet